SLC1A3 gene

solute carrier family 1 member 3

Normal Function

The *SLC1A3* gene provides instructions for making a protein called excitatory amino acid transporter 1 (EAAT1). EAAT1 transports a molecule called glutamate in the brain. Glutamate is one of several brain chemicals called neurotransmitters, which allow nerve cells (neurons) to communicate with one another. EAAT1 is found throughout the brain, but it is most abundant in the part of the brain that is connected to the spinal cord (the brainstem) and the region of the brain involved in coordinating movements (the cerebellum).

Neurotransmitters (such as glutamate) are released from neurons and relay signals to other cells by attaching to receptor proteins on neighboring neurons. After the neurotransmitters have had their effect, they detach from their receptors and must be cleared from the spaces between neurons. Researchers have determined that EAAT1 is one of several glutamate transporters that clear excess glutamate from these spaces. This process is carefully regulated to ensure that signals are transmitted accurately throughout the nervous system. The timely removal of glutamate is also necessary to prevent a buildup of this neurotransmitter between cells, which would be toxic to neurons.

Health Conditions Related to Genetic Changes

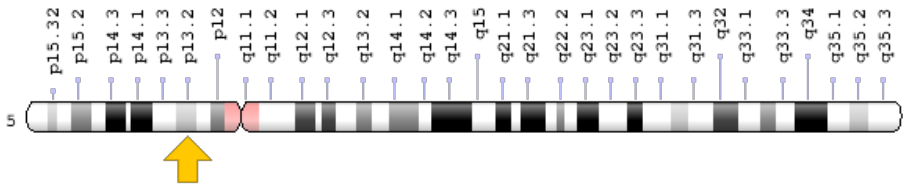
episodic ataxia

At least one mutation in the *SLC1A3* gene has been found to cause episodic ataxia type 6 (EA6). This mutation changes a single protein building block (amino acid) in the EAAT1 protein, replacing the amino acid proline with the amino acid arginine at position 290 (written as Pro290Arg). Research has shown that this genetic change likely impairs the ability of EAAT1 to remove glutamate from the spaces between neurons. The impaired uptake of glutamate may overexcite certain neurons in the brain, which disrupts normal communication between these cells. Although changes in signaling between neurons underlie the episodes of uncoordinated movement seen in people with episodic ataxia, it is unclear how altered glutamate transport causes the specific features of the condition.

Chromosomal Location

Cytogenetic Location: 5p13.2, which is the short (p) arm of chromosome 5 at position 13.2

Molecular Location: base pairs 36,606,355 to 36,688,334 on chromosome 5 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- EA6
- EAA1_HUMAN
- EAAT-1
- EAAT1
- Excitatory amino acid transporter 1
- FLJ25094
- GLAST
- GLAST1
- Glial high affinity glutamate transporter
- Glutamate/aspartate transporter, high affinity, sodium-dependent
- Sodium-dependent glutamate/aspartate transporter 1
- solute carrier family 1 (glial high affinity glutamate transporter), member 3
- Solute carrier family 1 member 3

Additional Information & Resources

Educational Resources

- Basic Neurochemistry (sixth edition, 1998): Glutamate Transporters
  https://www.ncbi.nlm.nih.gov/books/NBK27931/
- Neuromuscular Disease Center, Washington University: Episodic ataxia type 6
  http://neuromuscular.wustl.edu/ataxia/domatax.html#ea6

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28SLC1A3%5BTIAB%5D%29+OR+%28%28EAAT1%5BTIAB%5D%29+OR+%28GLAST%5BTIAB%5D%29+OR+%28GLAST1%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- SOLUTE CARRIER FAMILY 1 (GLIAL HIGH AFFINITY GLUTAMATE TRANSPORTER), MEMBER 3
  http://omim.org/entry/600111

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_SLC1A3.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=SLC1A3%5Bgene%5D
- HGNC Gene Family: Solute carriers
  http://www.genenames.org/cgi-bin/genefamilies/set/752
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=10941
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/6507
- UniProt
  http://www.uniprot.org/uniprot/P43003

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